CASE

- Parents of a 5-Year-old boy present to a clinic of a tertiary care hospital with complaints that the child has not yet started walking unsupported or speaking, & has looks very different from his siblings. On asking parents also report delayed sitting, & that he was also a slow learner. He could not feed himself & was not yet toilet trained also. He was born about 10 year later than their last child when mother’s age was 45 years.

- All other family members were normal.

ON EXAMINATION

- He appeared shorter for his age and plump. He had a small head; tongue appeared larger because it appeared to be protruding out of the mouth all the time. He had depreessed nasal bridge and small ears with folded ear pinna. He could barely talk but could communicate with gestures. Hands were small with fifth finger bent towards the fourth and there was only single palmer crease.

EEG: Recurrent Seizure discharges.

• ECG: Prolonged QRS complex.
• Blood sugar: 205mg/dl
• All other investigations were normal.

Q1 : What is this condition?
Down’s syndrome TRISOMY 21(Down syndrome is a genetic condition in which a person has 47 chromosomes instead of the usual 46.) Trisomy 21 (47, XX, +21) is caused by a meiotic nondisjunction event. With non-disjunction, a gamete (i.e., a sperm or egg cell) is produced with an extra copy of chromosome 21; the gamete thus has 24 chromosomes. When combined with a normal gamete from the other parent, the embryo now has 47 chromosomes, with three copies of chromosome 21.

Q2: Why other siblings are not affected?
- Not genetically transferred

Q3 : Which part(s) of the cell is involved, how and when does this disorder happen?

Q 4: What Organs/systems are involved in this condition?
- Nearly all systems of the body are involved in it.
  Incidence: 1 per 660 live birth, closely related to maternal age
  o Mother age<30 year risk:1 per 5000
  o Mother age>35 year risk:1 per 250

Q5: Name some other conditions caused by Abnormalities occurring at this level.

Q6: Can you explain the abnormal results of Investigations on basis of abnormal functioning of single part of a cell?

Q7: How does this condition affect the life expectancy & functional status of a person?

Q8: What social issues are faced by the child & the parents?
Q9: What is the earliest time when this condition can be diagnosed & what ethical issues are associated with diagnosis at this time?
- Many of the common physical features of Down syndrome may also appear in people with a standard set of chromosomes, including microgenia (an abnormally small chin), an unusually round face, macroglossia (protruding or oversized tongue), an almond shape to the eyes caused by an epicanthic fold of the eyelid, upslanting palpebral fissures (the separation between the upper and lower eyelids), shorter limbs.
- A single transverse palmar crease (a single instead of a double crease across one or both palms, also called the Simian crease), poor muscle tone and a larger than normal space between the big and second toes. Health concerns for individuals with Down syndrome include a higher risk for congenital heart defects, gastroesophageal reflux disease, recurrent ear infections, obstructive sleep apnea, and thyroid dysfunctions.

Flattened face
Mental retardation
Congenital heart disease: 50% Infection is common
Premature aging, Life expectancy up to 45 years. Musculoskeletal problems Physical development is often slower than normal. Most children with Down syndrome never reach their average adult height. Children may also have delayed mental and social development. Common problems may include:
- Impulsive behaviour
- Poor judgment
- Down syndrome in a fetus can be identified through chorionic villus sampling or amniocentesis during pregnancy, or in a baby at birth.
- It is statistically more common with older parents due to increased mutagenic exposures upon some older parents' reproductive cells.
- The average IQ of children with Down syndrome is around 50, compared to normal children with an IQ of 100. A small number have a severe to high degree of intellectual disability

Down Syndrome
- **Enabling Objective:**
  - By the end of the Module II, student will be able to:
  - Correlate the changes in structure and function of the different cell parts in health and in disease states.
  - Name few clinical common conditions associated with abnormal structure and function of different cell parts.

Learning Objectives:
- Structure, function and different parts of cell.
  Importance of cell membrane in regulating various function and defects in disease states.
  Importance of cell nucleus in defining cell function and determining phenotype and genotype of the species.
- Structure, number and types of chromosomes.
- Effects of Chromosomal anomalies on gross appearance and physiological functions of human body
- Clinical feature of this syndrome and name it.
  Social and ethical issues associated with this syndrome.
  Other syndromes of numerical and structural chromosomal abnormalities.